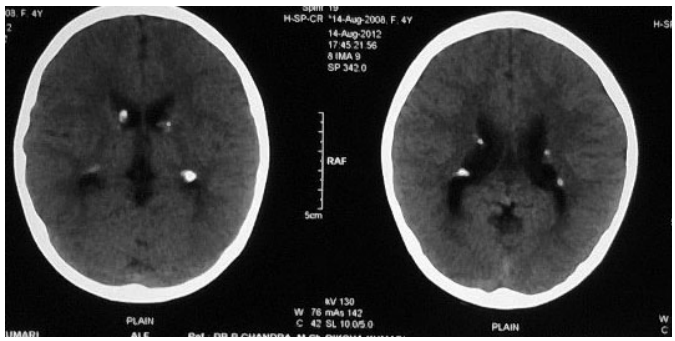
SPOT DIAGNOSIS (IMAGE GALLERY)



EPILEPSY WITH RENAL CYSTS

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A 4 year old girl suffering from seizures since 2 years of age on sodium valproate presented with pain in abdomen. Milestones were normal. On examination on abdominal system examination, bilaterally ballotable mass was palpable. Few hypomelanotic patches were present over the chest and trunk. Over the lumbosacral area on the back there were raised skin lesions like onion peel. Ultrasound {USG} abdomen was suggestive of multiple renal cysts bilaterally. CT brain showed multiple subependymal calcifications hanging from the walls of lateral ventricles. Following are the CT scan pictures.

What is the diagnosis?

Tuberous sclerosis complex. It is an autosomal dominant condition. The two genes responsible are TSC1 on chromosome 9 and TSC2 on chromosome 16. TSC 1 codes for hamartin and TSC 2 codes for tuberin. Loss of either of the proteins results in the formation of numerous benign tumors called hamartomas throughout the body involving skin, brain, heart, kidneys, liver and lungs. (1) Clinical and radiographic features of tuberous sclerosis complex have now been divided into major and minor categories based on the apparent degree of specificity for tuberous sclerosis complex of each feature. Definitive diagnosis requires at least 2 major or 1 major and 2 minor features. (2) Our patient presented with abdominal pain and had bilaterally palpable mass per abdomen which turned out to be multiple renal cysts in both the kidneys. She had history of convulsions since 2 years of age, controlled on sodium valproate. She was fulfilling 3 major criteria – subependymal nodules along the walls of lateral ventricle, more than 3 hypomelanotic patches and shagreen patch over the back. She had 1 minor criteria as well i.e. bilateral multiple renal cysts. (3)

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